

Delayed recognition of perineal ectopic testis in a pediatric patient: diagnostic and surgical implications of a rare cryptorchidism variant

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ABSTRACT

Cryptorchidism is one of the most prevalent congenital urogenital anomalies in the paediatric male population. Ectopic testis cases are less frequent, with perineal localization being the rarest form. A six-year-old patient was referred to the paediatric urology outpatient clinic following the failure to palpate the right testis during a routine physical examination in primary care. The evaluation process revealed the presence of a perineal ectopic testis, a diagnosis that was subsequently confirmed through ultrasonography. The testis was surgically mobilised, placed in a dartos pouch, and fixed in the scrotum. Perineal ectopic testis is a diagnostically challenging and rare variant. Even though the diagnosis was made at an older age in this case, the surgical procedure resulted in anatomically successful outcomes. The utilisation of systematic genital examinations within primary healthcare settings facilitates the timely identification and subsequent referral of cases exhibiting similar characteristics. Early diagnosis is of pivotal importance in preserving testicular function. It is important to note that even in cases where the onset of symptoms is delayed, the implementation of appropriate surgical intervention can result in favourable outcomes. The quality of primary care follow-up is fundamental to this process.

Keywords: Cryptorchidism, perineal testis, pediatric urology, scrotal orchidopexy, primary care, delayed diagnosis

INTRODUCTION

Cryptorchidism is among the most frequently reported congenital anomalies of the male genital system. It is characterised by the failure of the testis to complete its normal descent.¹ While most cases involve the testis being located in the inguinal canal or abdominal cavity, a small subset presents with ectopic testicular placement outside the usual descent path.² In the context of ectopic locations, the perineal region is the least prevalent, with an occurrence rate of less than 1 in 10,000 cases.³

Perineal ectopic testis is a rare variant that poses significant diagnostic challenges. In instances where the testis is not palpable in the standard locations, the possibility of ectopic placement must be considered. The failure to recognise such variants in a timely manner may result in complications, including testicular atrophy, subfertility, and malignant transformation.^{1,4}

Primary healthcare settings represent the fundamental framework for paediatric monitoring, and genital examination should be considered a vital component of routine assessments. Systematic physical examinations have

been shown to facilitate the early detection and referral of undescended or ectopic testis cases.⁵ The present report details a case of delayed diagnosis of perineal ectopic testis, addressing the diagnostic process, surgical approach, and the critical role of primary care from an academic perspective.

CASE

A six-year-old male patient was referred to a specialist in paediatric urology following a routine physical examination in primary care that revealed an empty right hemiscrotum. Physical examination revealed the presence of the urethral meatus at the coronal level, in addition to a dorsally hooded prepuce. Surgery was planned for hypospadias repair in the second session. The left testis was found to be in its normal position, exhibiting the expected dimensions and consistency that are characteristic of its age. The right hemiscrotum was found to be hypoplastic, with only minimal fluid present (Figure 1). It was not possible to palpate the right testis in the inguinal canal.



Figure 1. Preoperative perineal appearance

Upon extending the palpation area, a mobile, soft mass was detected approximately 1.5 cm inferior to the scrotum in the right perineal region, overlying muscle tissue. Ultrasonography confirmed the absence of the right testis in the scrotum and identified the perineal mass as testicular tissue.

Surgical exploration was performed through a transverse scrotal incision. After the traversal of the scrotal layers, the dissection was deepened towards the perineal area, where the testis was located. The adhesions to the surrounding tissues were meticulously released. The testis, exhibiting normal consistency and displaying a slightly reduced size in comparison with the contralateral side, was mobilised (**Figure 2**). The epididymis was then found to be separated. There was no tension when the testicle was placed in the dartos sac and fixed to the scrotum. (**Figure 3**). The postoperative course was uneventful, with no complications observed.

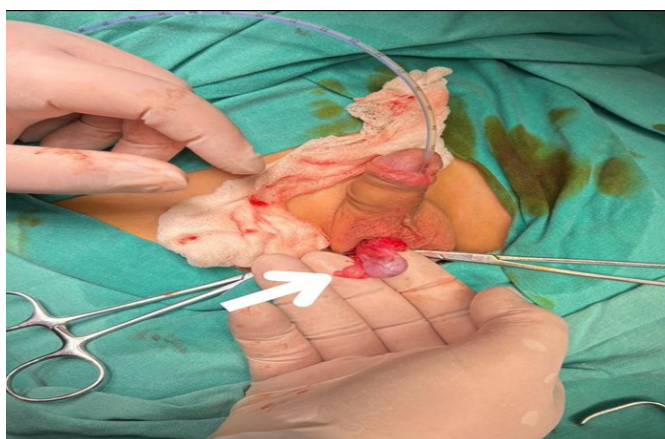


Figure 2. Dimensions of the testis



Figure 3. Postoperative single incision appearance

DISCUSSION

Perineal ectopic testis is characterised by abnormal testicular descent, deviating from the physiological route and settling in the perineal area, making it an extremely rare form of cryptorchidism.⁶ While ectopic testis accounts for approximately 5-10% of all undescended testes, perineal placement is the least common variant within this group. Diagnosis is frequently challenging, as the testis is not located in the conventional palpation zones and may be misinterpreted as testicular agenesis or intra-abdominal testis.⁷ Atypical anatomical positioning necessitates a careful and systematic approach during physical examination and surgical planning. This case, despite presenting typical findings associated with cryptorchidism, is diagnostically significant due to its perineal location, which represents a variant that has been rarely reported in the literature. Despite the diagnosis being delayed, the correct localisation was achieved via physical examination and successful scrotal fixation was performed through a minimally invasive approach, rendering the case clinically relevant.

In primary healthcare settings, routine genital examinations during childhood follow-ups are crucial for the early recognition of urogenital anomalies, such as undescended or ectopic testes. In this particular instance, the diagnosis was made at a relatively late stage, at six years of age. This suggests that earlier physical evaluations may have been either overlooked or not carried out in a sufficiently systematic manner. This diagnostic delay has the potential to increase the risk of complications. Consequently, structured, documented, and auditable genital examinations should be standard practice in paediatric follow-ups. Furthermore, the implementation of continuous professional development for healthcare providers, in conjunction with public education campaigns for parents, has the potential to enhance clinical awareness and societal consciousness.

Although there are different opinions in the literature about the use of routine ultrasonography in the diagnosis of undescended testis, it can be a valuable aid in cases with suspected ectopic testis.¹ Ultrasonography is a valuable tool in a range of medical contexts, including the identification of anatomical positioning, assessment of vascularization, and guidance for surgical planning. However, it should be noted that the sensitivity of the test varies, especially in cases where it is performed in an abdominal or atypical location.⁸ Consequently, if clinical suspicion remains high despite inconclusive imaging, exploratory surgery should not be postponed.⁹ In the presented case, the testis was not located in the inguinal canal but was identified in the perineal region via extended palpation and confirmed with ultrasonography.

From a surgical standpoint, the ability to localise the problem with precision through physical examination and targeted imaging has been shown to prevent unnecessary laparoscopic or inguinal explorations due to diagnostic uncertainty. A solitary scrotal incision facilitated the mobilisation of the testis from the perineum, achieving minimal tissue trauma. It was observed that the vascular structures were preserved, and that the testis was tension-free when placed in the dartos pouch. The length of the cord was found to be adequate, thus



enabling the procedure to be completed in a single session without the need for additional surgical intervention. As documented in the extant literature, surgical planning in ectopic testis cases should be based on a detailed physical assessment and meticulous localization, with a view to minimising unnecessary invasive procedures, operative time, costs, and patient morbidity, while optimising anatomical and functional outcomes.¹⁰ A systematic examination is fundamental to the management of patients in whom the testis is not found in the scrotum or inguinal canal, with the aim of excluding perineal and other ectopic locations.

CONCLUSION

Perineal ectopic testis is a rare form of cryptorchidism that poses significant diagnostic challenges due to its atypical localization. This case demonstrates that, despite delayed diagnosis, favourable anatomical and functional outcomes are attainable with accurate localization and appropriate surgical management. Standardised genital examinations in primary care remain essential for the early detection and prevention of long-term complications.

ETHICAL DECLARATIONS

Informed Consent

Informed consent was obtained from the legal guardians of the pediatric patient(s) described in this report. Where developmentally appropriate, assent was also sought from the child. The inclusion of vulnerable populations in this study adhered to national and international ethical guidelines. Extra care was taken to ensure voluntary participation, understanding, and protection of participant dignity and autonomy.

Peer Review Process

This report underwent external peer review.

Conflict of Interest

The authors declare no conflicts of interest.

Financial Disclosure

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Author Contributions

Concept: K.N., T.H.; Design: K.N., T.H.; Control: K.N., T.H.; Resources: K.N., T.H.; Materials: K.N.; Data Collection and/or Processing: K.N.; Analysis and/or Interpretation: K.N.; Literature Review: T.H.; Writing the Article: K.N., T.H.; Critical Review: K.N., T.H.

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